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ACCOUNTING FOR CONGENITAL MALFORMATIONS IN NORTH CAROLINA

by

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ABSTRACT

North Carolina death certificate data were utilized to provide a general description of mortality due to congenital malformations. A similar examination of the incidence of malformations among North Carolina births began with an assessment of the completeness of reporting malformations on birth certificates. Birth certificate data for 1980 were compared with newborn records from the 1980 hospital discharge data. It was found that North Carolina birth certificates reflected only about 20 percent of the congenital anomalies cited on the hospital discharge records of newborns in 1980. This suggests a strong need for developing a more complete system of reporting malformations among North Carolina's live births.

Introduction

Congenital malformations are a focus of concern in the public health and medical communities due in part to their ranking as a leading cause of infant mortality. Such anomalies were listed as the underlying cause for the deaths of 335 North Carolinians in 1982. Over 70 percent of these were persons under one year of age.

Beyond the mortality issue there are also ongoing medical, social, and economic implications associated with birth defects in relation to childhood morbidity. These long-term effects stem from the medical and physical handicaps which frequently accompany serious anomalies.

For the purposes of the present study, a congenital malformation is defined as "an abnormal condition that exists at (or usually before) birth and results from a hereditary defect, an environmental agent, or the interaction of both." (5) This research has been confined to analysis of codes 740-759 of the International Classification of Diseases (9th revision) and excludes birth injuries and other complications of pregnancy.

The causes of congenital anomalies are of increasing epidemiologic interest, especially with the introduction of numerous new drugs and chemicals into the environment, many of which may have teratogenic potential. Effective research into the causes of birth defects necessitates accurate reporting of their occurrence. While a reasonably good picture of deaths due to these causes can be obtained using data supplied on death certificates, there is considerable question as to whether official birth records provide an adequate accounting of the incidence of malformations among live births. (5,9)

It is this latter question which is a primary concern in this paper. The purposes of this report are to describe the reported incidence of congenital malformations among the births and deaths of North Carolina residents, examine the completeness of reporting anomalies on birth certificates, and assess the usefulness of these data for monitoring and epidemiologic research.

Methods

Examination of mortality due to congenital malformations involved the use of death certificate data for which the underlying cause of death was listed as 740-759 according to the 9th Revision of the International Classification of Diseases (ICD-9) as published by the World Health Organization, and adopted for use in 1979. Data from years coded by ICDA-8th Revision were also utilized, but codes 740-759 were not changed in the 9th Revision.

A primary purpose of this research is to determine the completeness of reporting congenital malformations on birth certificates in North Carolina. Therefore, data from birth certificates were compared to newborn records included in 1980 hospital discharge data files. Patient discharge summaries for over 90 percent of North Carolina general hospital inpatients were collected in computerized form for the calendar year 1980. To enhance comparability, birth certificate records were utilized only if the births occurred in hospitals for which the discharge data were available. Records for non-residents of North Carolina were deleted from both sets of data, leaving only resident live births which occurred in selected North Carolina general hospitals. Thus the study covered 86% of all 1980 North Carolina resident live births.

Congenital malformations on birth certificates are coded according to the ICD. Up to two specific conditions may be coded, and there is an indicator to note the existence of additional conditions. There are up to seven diagnoses on the 1980 hospital discharge records, coded using the Clinical Modification of ICD-9. This modification allows for coding a fifth digit, which gives information specific to the treatment of conditions by hospital personnel. The first four digits of the two adaptations are the same for congenital malformations. It was thought that the availability of up to seven specific codes for each hospital discharge record and only two for each birth record might adversely affect comparisons between these data sets. For each data set, however, only about 2.6 percent of the records had three or more malformations. Thus, any problems resulting from the availability of additional codes on discharge records were minimal.

Another set of data utilized was compiled by the Centers for Disease Control (CDC) in Atlanta, Georgia, as a part of their Birth Defects Monitoring Program.(1) These data include live births and fetal deaths occurring in selected North Carolina hospitals. The events recorded were not limited to North Carolina residents, nor were the hospitals monitored necessarily the same as those represented by the 1980 hospital discharge data. Thus, direct comparison of numbers of events between these data sets was not possible. The CDC data, however, included a rate of the number of malformations cited to the number of deliveries recorded in their selected hospitals. This rate was calculated for the hospital discharge data and comparisons were made for 1980.

To more clearly approximate the results obtained from the CDC analysis, the hospital discharge data were tabulated to include newborns and fetal deaths that occurred in participating hospitals regardless of residence status. Further, the CDC data were coded according to the Birth Defects Monitoring Program (BDMP) coding scheme which combined some categories and also incorporated some additional codes. The discharge data were recoded to match the BDMP scheme. A list of the code equivalents is provided in Appendix A. Only selected malformations were examined due to very small numbers in some categories.

Mortality Due to Congenital Malformations

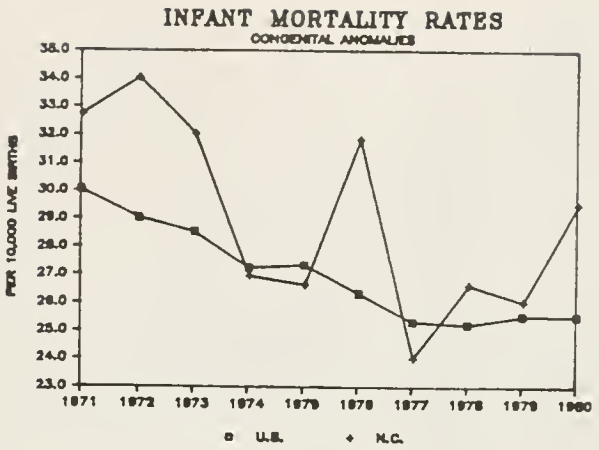
Although congenital malformations accounted for less than 1 percent of all 1982 deaths to North Carolina residents, 20 percent of all infant deaths (under 1 year of age) were attributed to such anomalies, as were about 12 percent of deaths among children 1 to 4 years of age. Table 1 shows the five leading causes of mortality for North Carolina infants (under one year of age) and children 1 to 4 years old in 1982.

Among congenital malformations, the specific conditions most often reported for decedents through four years old were: congenital anomalies of the heart (ICD 745, 746) and circulatory system (ICD 747), congenital anomalies of the respiratory system (ICD 748), hydrocephalus and other anomalies of the nervous system (ICD 742), unspecified and multiple anomalies (ICD 759), and anencephalus (ICD 740).

Table 1
Leading Causes of Mortality for Infants and Children 1 to 4 Years Old
North Carolina 1982

Infants			1-to-4 year-olds		
Condition	Number of Deaths	Percent Of Total	Condition	Number of Deaths	Percent Of Total
Anoxia and Hypoxia	240	20.4	Accidents	78	43.3
Congenital Malformations	235	20.0	Congenital Malformations	22	12.2
Sudden Infant Death Syndrome	144	12.2	Homicide	12	6.7
Immaturity	135	11.5	Cancer	11	6.1
Conditions of mother and difficult labor	83	7.1	Pneumonia & Influenza/Meningitis	7	3.9
All Causes	1,176	100.0	All Causes	180	100.0

Since the majority of deaths from congenital anomalies occurs among infants, mortality rates are frequently calculated like crude infant mortality rates, i.e., using the number of live births in the denominator. The accompanying figure depicts infant deaths due to congenital malformations per 10,000 live births for the United States and North Carolina from 1971 through 1980 (1980 data are the latest available for the nation). As this graph indicates, rates for the state were erratic during this period, fluctuating from a high of 34.0 in 1972 to a low of 24.0 in 1977 and back up to 29.5 in 1980 (the 1982 rate for N.C. was 27.4). Rates for the United States showed a more consistent decline followed by a leveling-off at 25.5 in 1979 and 1980.



Incidence of Congenital Malformations Among North Carolina Births

There were 749 congenital anomalies listed on birth certificates of North Carolina residents in 1982. This number would indicate that less than 1 percent of the state's 85,908 liveborns had malformations. However, there is growing concern that many malformations are not recorded on certificates of birth, in part because some anomalies are not readily identifiable at birth. (9)

In an attempt to discover the degree of underreporting of malformations on birth certificates in North Carolina, a comparison was made of birth certificate data and hospital discharge records for newborns for calendar year 1980. Table 2 shows the rates for each malformation code 740-759 per 10,000 live births for the two sets of data. Again, both the birth certificate and hospital data are for North Carolina resident births occurring in the same set of hospitals, so the numbers shown in the first columns should be roughly comparable. As indicated by these rates, birth certificates on the whole revealed only about 20 percent of the anomalies diagnosed on discharge records. This discrepancy is reiterated by the ratio of the number of malformations on birth certificates to the number on discharge records, adjusted for the number of births represented in each data set. These ratios are also included in Table 2 and show that, of the anomalies most likely to cause death, only anencephaly was well reported on the birth certificates (the ratio was 1.31 mentions of anencephaly on birth records for every one mention on discharge records). Spina bifida (ICD 741) and cleft palate and lip (ICD 749) were also relatively well reported, with ratios of 0.70 and 0.69 respectively.

Table 2
Frequencies of Malformations for Births Occurring in Selected N.C. Hospitals by ICD Code, with Malformations per 10,000 Live Births

ICD	Condition	Birth Certificate Data		Hospital Discharge Data		Ratio*
		Number	Rate†	Number	Rate†	
740	Anencephalus	16	2.21	12	1.69	1.31
741	Spina bifida	40	5.51	56	7.88	0.70
742	Other anomalies of nervous system	18	2.48	80	11.26	0.22
743	Anomalies of eye	3	0.41	36	5.06	0.08
744	Ear, face and neck	17	2.35	131	18.43	0.13
745	Bulbus cordis anomalies	3	0.41	127	17.87	0.02
746	Other anomalies of heart	13	1.79	196	27.57	0.06
747	Other circulatory system	5	0.69	280	39.39	0.02
748	Respiratory anomalies	6	0.83	49	6.89	0.12
749	Cleft palate and lip	63	8.70	89	12.52	0.69
750	Other upper alimentary tract	12	1.66	79	11.11	0.15
751	Other digestive system	11	1.52	71	9.99	0.15
752	Genital organ anomalies	42	5.80	448	63.03	0.09
754	Musculoskeletal deformities	54	7.45	369	51.91	0.14
755	Other limb anomalies	187	25.81	501	70.48	0.37
756	Other musculoskeletal	27	3.73	137	19.27	0.19
757	Anomalies of integument	38	5.25	198	27.86	0.19
758	Chromosoman anomalies	22	3.04	76	10.69	0.28
759	Multiple and unspecified	36	4.97	75	10.55	0.47
	Total Malformations	620	85.58	3050	429.10	0.20
	Births with Malformations	578	79.78	2752	387.17	0.21
	Total Live Births	72,449		71,079		

*Ratio = $\frac{\text{number of malformations (birth certificate data)/number live births}}{\text{number of malformations (discharge data)/number live births}}$

†Rate = $\frac{\text{number malformations}}{\text{number live births}}$ x 10,000

Congenital Malformations by Race and Sex

Although there are a number of pertinent demographic variables of interest included on birth certificates, analysis of these in relation to malformations could prove invalid given their incomplete reporting. Thus, further description of North Carolina births with congenital anomalies was limited to their rates of occurrence by race and sex as recorded in the 1980 hospital discharge data (Table 3).

The data in Table 3 reveal a higher incidence of malformations among males than females. However, this difference results primarily from the higher proportion of males with genital anomalies (the ratio of males to females for anomalies of the genital organs—ICD 752—was 14.81). The ratio of total malformations among males to total malformations among females was 1.24. However, eliminating anomalies of the genital organs from the totals results in a male-to-female malformation ratio of 0.93. Also shown in Table 3 are the ratios of malformations for white to nonwhite births. While nonwhites were reported to have a higher incidence of total malformations (white to nonwhite ratio of 0.83), white infants had a higher incidence reported for several specific malformation categories. Among these, the most pronounced discrepancy was for cleft lip and palate (white to nonwhite ratio of 2.72). Overall, the rate of total malformations per 10,000 live births was highest for nonwhite males (479.8) and lowest for white females (319.0).

Table 3

Rates* of Selected Malformation Categories by Race and Sex from 1980 Hospital Discharge Data, with Male to Female and White to Nonwhite Ratios†

	White			Nonwhite			Total††			Ratio†	
Condition	Male	Female	Total	Male	Female	Total	Male	Female	Total	White to Nonwhite	Male to Female
Anomalies of Nervous System (740-742)	25.3	22.0	23.7	11.6	18.1	14.8	20.8	20.5	20.7	1.60	1.01
Anomalies of the Heart (745-746)	45.4	44.4	43.9	46.5	47.0	46.8	45.5	45.1	45.3	0.96	1.01
Other Circulatory System Anomalies (747)	41.4	28.9	35.4	51.9	42.5	47.2	44.4	33.2	39.0	0.75	1.34
Cleft Palate and lip (749)	18.5	12.9	15.8	7.2‡	4.5‡	5.8‡	14.8	10.1	12.5	2.72	1.47
Anomalies of the Genital Organs (752)	110.9	7.3‡	60.9	126.2	9.0‡	67.9	115.5	7.8	63.0	0.90	
Musculoskeletal, including hip dislocation (754)	39.8	77.6	58.0	35.8	41.6	38.7	39.2	65.3	51.9	1.50	
Reduction and other limb deformities (755)	44.6	43.1	43.9	127.1	130.2	128.6	69.7	70.8	70.2	0.34	0.98
Chromosomal anomalies including Down's Syndrome (758)	10.0	12.9	11.4	7.2‡	10.9‡	9.0	9.3	12.1	10.7	1.27	0.77
Other unspecified and multiple anomalies (759)	12.1	11.6	11.9	7.2‡	7.2‡	7.2‡	10.4	10.1	10.3	1.65	1.03
Total anomalies (740-759)	404.7	319.0	363.3	479.8	397.0	438.6	426.0	343.2	38.56	0.83	1.24

* Rate = $\frac{\text{number of malformations}}{\text{number of live births}}$ x 10,000 (race-and sex-specific)

† Ratio = $\frac{\text{number of malformations/number of births (white or male)}}{\text{number of malformations/number of births (nonwhite or female)}}$

†† Total includes cases for which race and/or sex were not stated.

‡ Numerator under 20

Trends in Congenital Malformations

Rates per 10,000 total deliveries (live births plus fetal deaths) for selected congenital malformations are shown in Table 4 for the hospital discharge and CDC data for 1980. Although the total numbers of deliveries reported for the two data sets were different (75,777 for hospital discharge data and 44,840 for the CDC data), the rates for selected conditions were usually very similar. Thus, the CDC data were examined for trends over time.

The data presented in Table 5 are from the CDC's Birth Defects Monitoring Program (BDMP). These data show rates of malformations to total deliveries in selected hospitals for the United States and North Carolina for 1970-82 and each year 1970, 1975, and 1982, with the percentage changes in rates from 1970 to 1982. Overall, the state's total rates (the cumulative number of anomalies for 1970 through 1982 divided by the total number of deliveries for these years and multiplied by 10,000 for each malformation category) were lower than those for the entire United States. Exceptions are congenital anomalies of the nervous system (BDMP Codes 125—total congenital anomalies of the nervous system, 45—anencephaly, and 56—spina bifida without anencephaly), but North Carolina's rates for these conditions decreased more dramatically during the period.

Table 4

Comparison of 1980 Hospital Discharge and CDC Data for Selected Congenital Malformation Rates*

BDMP	Code/Condition	Discharge Data	CDC Data	Ratio†
45	Anencephaly	3.7	4.0	0.93
56	Spina bifida without anencephaly	7.9	8.0	0.99
65	Hydrocephalus without spina bifida	5.5	2.5	2.20
125	Total congenital anomalies of the nervous system	23.0	18.5	1.24
200	Ventricular septal defect	11.5	9.1	1.26
260	Patent ductus arteriosus	24.4	16.5	1.48
329	Cleft lip with or without cleft palate	7.9	7.8	1.01
393	Rectal atresia and stenosis	3.0	2.7	1.11
413	Hypospadias and epispadias	24.7	23.0	1.07
472	Clubfoot without CNS defect	22.6	18.5	1.22
483	Reduction deformities	4.2	2.7	1.56
500	Hip dislocation without CNS defect	20.3	21.4	0.95
626	Down's Syndrome	7.1	6.5	1.09
943	RH Hemolytic disease	10.4	9.1	1.14

Totals were not available for the CDC data.

* Rate = (malformations/live births plus stillbirths) x 10,000

† Ratio = $\frac{\text{malformations/live births plus still births (discharge data)}}{\text{malformations/live births plus still births (CDC data)}}$

Several categories of malformations in the state warrant special concern. Among these, the N. C. rate for cleft lip with or without cleft palate (BDMP 329) fluctuated throughout the 1970-1982 period and showed an overall increase, in sharp contrast to the consistent decline in this rate for the United States. Also, Down's Syndrome (BDMP 626) increased after similar fluctuations, while the U.S. rate for Down's Syndrome showed a small decrease from 1970 to 1982. Dramatic increases were noted both in North Carolina and nationwide for certain anomalies of the heart (BDMP Codes 200-ventricular septal defect, and 260-patent ductus arteriosus) and for congenital hip dislocation without central nervous system defect (BDMP 500).

It should be noted that physician diagnosis and coding practices may have changed to some extent over this period, affecting the trends shown in Table 5. Also, caution should be maintained in interpreting single-year rates for specific conditions in the North Carolina data. This is due to small numbers in the numerators (under 20 events) for some of the rates.

Table 5

Rates* of Selected Malformations from CDC Data for 1970-1982 With Single-Year Rates for 1970, 1975, and 1982 and Percentage Changes from 1970 to 1982: U.S. and N.C.

		United States					North Carolina				
BDMP	Code/Condition	Total 1970-82	1970	1975	1982	% Change 1970-82	Total 1970-82	1970	1975	1982	% Change 1970-82
45	Anencephaly	4.2	5.6	4.3	3.3	- 41.1	4.8	7.6	5.1	2.2	- 71.1
56	Spina bifida w/o anencephaly	5.8	7.2	5.5	4.6	- 36.1	8.0	11.4	8.1	5.8	- 49.1
65	Hydrocephalus w/o spina bifida	4.5	4.8	3.9	5.0	+ 4.2	4.1	5.0	3 0‡	5.4	+ 8.0
125	Total anomalies of nervous system	18.8	21.8	18.3	17.7	- 18.8	21.2	29.6	20.7	18.0	- 39.2
200	Ventricular septal defect	8.6	4.0	7.8	14.7	+ 267.5	6.4	3.2‡	7.1	10.3	+ 221.9
260	Patent ductus arteriosus	12.5	3.5	10.5	25.4	+ 625.7	10.1	3.6‡	10.4	15.5	+ 330.6
329	Cleft lip with or w/o cleft palate	9.1	9.7	9.5	8.8	- 9.3	8.2	8.6	7.3	9.0	+ 4.7
393	Rectal atresia and stenosis	3.3	3.5	3.2	2.9	- 17.1	3.0	4.0	3.7‡	3.8‡	- 5.0
413	Hypospadias and epispadias	23.8	20.2	23.5	27.0	+ 33.7	20.1	15.1	22.9	19.5	+ 29.1
472	Clubfoot w/o CN5 defect	25.8	25.6	23.3	24.5	- 4.3	18.2	23.5	17.3	18.4	- 21.7
483	Reduction deformities	3.4	3.3	3.2	3.5	+ 6.1	3.1	3.4‡	3.2‡	4.7	+ 38.2
500	Hip dislocation w/o CN5 defect	21.3	6.8	20.8	27.0	+ 297.1	12.9	3.4‡	12.8	13.7	+ 302.9
626	Down's syndrome	8.0	8.1	8.6	7.9	- 2.6	5.6	4.6	5.9	5.8	+ 26.1
943	RH Hemolytic disease	23.2	44.9	20.4	15.6	- 65.3	12.9	21.0	14.4	6.7	- 68.1

* Rate = malformations/(live births plus fetal deaths) x 10,000

‡ Numerator under 20

Summary and Future Directions

Congenital malformations were the second leading cause of infant mortality in North Carolina in 1982. While the rate of these deaths per 10,000 live births declined from 32.5 in 1971 to 27.4 in 1982 (a 15.7 percent decrease), this decline was not continuous from year to year and state rates remained higher than those for the U.S. as a whole. Congenital anomalies as a cause of death can be examined utilizing data from death certificates, but such analysis does not provide an assessment of anomalies among the living.

A severe limitation of research directed at identifying possible environmental causes of birth defects has been the lack of a complete and accurate system for reporting malformations among live births. A major question for this study was whether North Carolina birth certificate data provide a complete accounting of the prevalence of congenital anomalies among the state's live births. The answer found here is that they do not.

The comparison of North Carolina birth certificate data with newborn data from 1980 hospital records revealed that, overall, only 20 percent of the malformations shown on the hospital records were reported on birth certificates. This finding is consistent with that of a similar study conducted by Seegmiller, et al. on Utah births, in which about 26 percent of the malformations recorded on discharge records were also included on birth certificates. (9)

While hospital records appear to be more inclusive of the actual incidence of malformations among births, there are limitations to the use of these data alone. Though more complete than the birth certificates, some underreporting is also likely on the hospital records. Further, such records often lack demographic variables which are pertinent to research. For example, analysis of congenital anomalies and other matters relating to infant mortality and morbidity are more thorough when data are available about the mother as well as the infant (e.g., age and education of mother, prior pregnancy history, and prenatal care information). While this type of information is available on birth certificates, the problem of underreporting of anomalies severely limits the use of birth certificates in isolation from other data sets.

What is needed, then, is a set of data which combines information from several sources to provide a more complete and thorough base from which monitoring and research activities can be conducted. The Missouri Center for Health Statistics has compiled such a data base for their state, utilizing birth certificates, hospital discharge data, death certificates, and Missouri Crippled Children's Service data. Records from each source were matched to form one data base of children under the age of two with congenital malformations. (4) As well as combining records from several sources to provide more information for each malformation, any one source may identify a malformation not captured by any of the others. The compilation of such a data base for North Carolina residents is currently under consideration. Completion of this project would necessitate cooperation from several agencies with relevant data. Most notably, continued access to hospital discharge records is imperative, since these records provide the most complete accounting of births with congenital malformations currently available.

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Appendix A

The conversion of the Clinical Modification of the ICD-9 coding scheme to correspond to that developed by the CDC's Birth Defects Monitoring Program produced the following code equivalents:

BDMP Code	ICD-9-CM (1st four digits)
45—Anencephalus	740.0-740.2
56—Spina bifida	741.0, 741.9 without 740.0-740.2
65—Hydrocephalus	742.3 without 741.0
125—Total congenital anomalies of the nervous system	740-740.2, 741.0, 741.9, 742.0-742.5, 742.8-742.9, 237.7, 331.3-331.4
200—Ventricular septal defect	745.3-745.4, 745.7
260—Patent ductus arteriosus	747.0
329—Cleft lip with or without cleft palate	749.1-749.2
393—Rectal atresia and stenosis	751.2
413—Hypospadius and epispadias	752.6
472—Clubfoot without central nervous system defect	754.5-754.7 without 740.0-740.2, 741.0, 741.9, 742.0
483—Reduction deformities	755.2-755.4
500—Hip dislocation without central nervous system defect	754.3 without 740.0-740.2, 741.0, 741.9,742.0
626—Down's syndrome	758.0
943—RH Hemolytic disease	773.0

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